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QUALITY OF LIFE OF PATIENTS WITH SHERESHEVSKY-TURNER SYNDROME

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Shereshevsky-Turner Syndrome (TSS) is a spectrum of phenotypic characteristics resulting from gene deficiencies on the second sex chromosome. According to experts, the syndrome occurs with a frequency of 1:2000-1:2500 newborn girls [15].

Shereshevsky -Turner syndrome is associated with short stature, delayed puberty, ovarian dysgenesis , hypergonadotropic hypogonadism , infertility, congenital heart disease (CHD), endocrine disorders (diabetes mellitus type 1 and 2, osteoporosis and autoimmune diseases). Morbidity and mortality in women with Turner syndrome is higher than in the general population. Despite the often-noticeable phenotype, the delay in diagnosis can be significant, and the median age of diagnosis is around 15 years [5].

The mechanisms that lead to these defects are not fully understood and are hidden by significant variability in both karyotype and phenotype, with no consistent correlation between them [6].

Approximately one third of girls with STS experience spontaneous puberty, but only half of them end puberty with menarche [3].

According to Jeż W et al [7] In some countries, there are about 8,000 women with Turner syndrome, of which 5,000 are over 18 years of age. SST significantly complicates the psychosocial functioning of patients, especially during the period of life when the feeling of physical attractiveness develops. Dissatisfaction with one's physical data contributes to a decrease in the quality of life.

Understanding some aspects of health-related quality of life based on the subjective assessments of patients with TSS will contribute to physicians' awareness of the needs of these women and thus allow better tailoring of the treatment process [4;7].

Cardiovascular anomalies are a major concern in both prenatal and postnatal periods. According to experts, CHD that are incompatible with life in 99% contribute to fetal death [12], while bicuspid aortic valve and aortic coarctation are more common complications of live births [9; 14].

It is possible that people with short stature, regardless of the cause, may have a lower quality of life (QoL) than people with normal stature [2].

Purpose of the study: to study the indicators of quality of life in patients with Shershevsky-Turner syndrome

The study included 6 9 females aged 16 to 23 years, the average age was 20.3±1.9 years (Me 20.0 years; IQR 19.0-21.0). The main group included 43 patients with STS , the mean age was 20.1±1.3 years (Me 20.0 years; IQR 19.0-21.0). The control group included 26 healthy individuals aged 20.5±2.6 years (Me 20.0 years; IQR 19.0-22.0). The groups were comparable in terms of age (p=0.40) and BMI (p=0.29).

Signs of STS were: short stature (97.7%), primary amenorrhea (90.7%), sexual infantilism (79.1%), wide chest (62.8%), X-shaped curvature of the legs (58.1 %), skin folds on the neck (55.8%), arterial hypertension (37.2%), hearing loss (25.6%). 7 (16.3%) patients were married, but subsequently divorced due to diagnosed infertility. The mean height of the patients was 136±5.1 cm (Me 135.0 years; IQR 133.0-140.0) .

According to the results of karyotyping, 21 (48.8%) patients have the classic form of SST (monosomy X – 45, XO).

To assess the quality of life of patients, a short version of the health questionnaire (MOS 36 - Item Short - Form Health Survey - MOS SF-36). The 36 items of the questionnaire are grouped into eight scales: physical functioning (PF), role functioning (RP), body pain (BP), general health (GH), vitality (VT), social functioning (SF), role emotional functioning (RE) and mental health (MH).

Statistical analysis of the obtained data was carried out using Microsoft Excel 10 packages using the Data Analysis add-in and SPSS version 23. The data were checked for normal distribution using the Kolmagorov -Smirnov test. The results of analyzes are presented as median (Me) [interquartile range (IQR): 25th percentile; 75 percentile]. Differences were considered significant at $p< 0.05$.

Results and discussion

An analysis of the parameters of the physical component of health showed that with SST the integral indicator of the physical component of health (PCH) varied from 53.3 to 58.4 points with Me 55.8 points. These indicators are significantly lower than the control values (Me 76.6; IQR 71.4-80.5) (Table 1.).

Table 1.

Indicators of the physical health component of the quality of life of patients with TSS (according to SF-36), in points

Scales SF-36	**Control, n=2 6**	**WST, n= 43**
Physical functioning, **FF**	79.6±9.3	63.1±10.0*
Role functioning, **RF**	78.2±11.1	54.1±8.3*
Body pain, **B**	81.3±8.1	65.4±9.5*
General health, health **status**	82.3±11	53.5±9.4*
Physical component of health, **FKZ**	76.3±5.9	56.1±4.8*

Note: SST - Shershevsky -Turner syndrome ; * $p<0.0001$

Almost a third of the patients assessed role physical functioning (30.2% of respondents) and general health (32.6% of respondents) as low (less than 50 points).

A comparative analysis of the QoL components in the main and control groups revealed a sig-nificant decrease in FF (by 20.8%) compared to the control group, i.e. in patients with SST, the ability to perform various physical activities is significantly limited (Fig. 1a.).

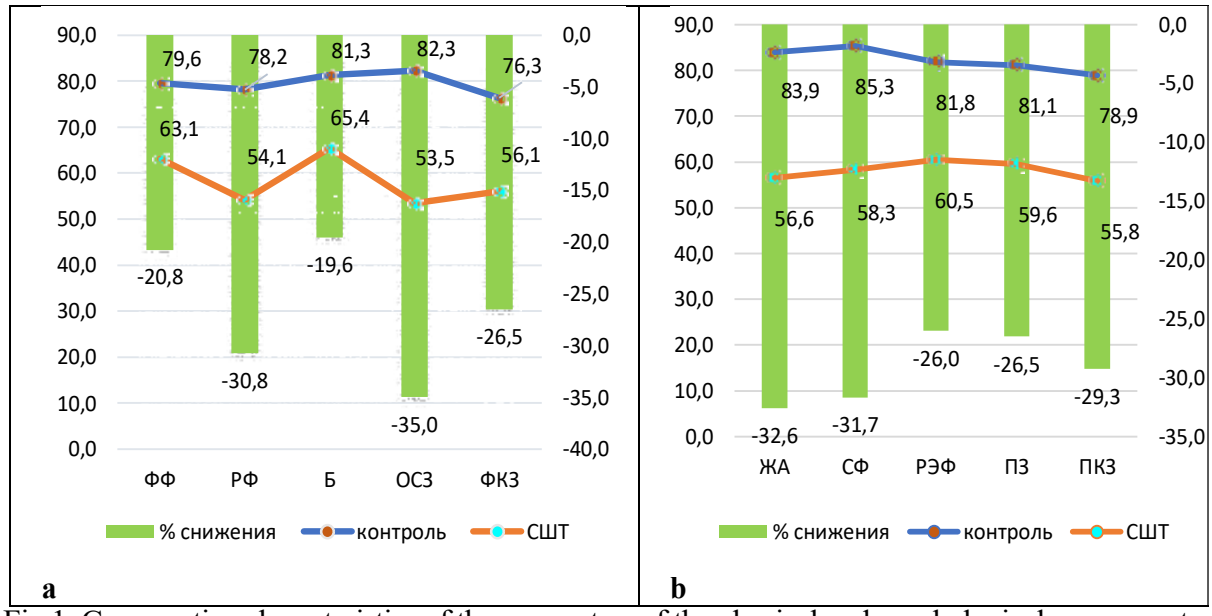

Fig.1. Comparative characteristics of the parameters of the physical and psychological components of the health of the quality of life of the SST and the control group.

SST has a negative impact on a person's daily role activities (work, performance of daily duties), there is a decrease in the RF component by 30.8%. A decrease in the bodily pain scale (B - by 19.6%) is mainly observed in persons with a history of arterial hypertension. All this led to a decrease in patients' subjective assessment of their health (PH - by 35.0%), as a result, the physical component of health was reduced by 26.5% .

A study in Norway showed that women with TSS were less likely to enjoy physical activity during childhood compared to controls [13; 17].

Thompson T. et al [17] reported that some of the patients with TSS admitted to feeling uncomfortable or inadequate when exercising with their peers. In the survey, social physical activity included weekend hikes with family, cycling with siblings, going to the gym with a friend, participating in a sports team, or attending a fitness class.

Amedro P. et al [1] found impaired quality of life in many areas, including physical functioning in both patients with TSS and their parents.

The integrated indicator of the psychological component of health (PKH) ranged from 52.3 to 59.5 points with Me 56.5 points. These indicators are significantly lower than the control values (Me 80.0; IQR 74.2-83.8) (Table 2.).

Table 2.

Indicators of the psychological component of health quality of life patients with SST (according to SF-36), in points

Scales SF-36	**Control, n=2 6**	**WST, n= 43**
Vital activity of **VA**	83.9±9.6	56.6±9.6*
Social functioning, **SF**	85.3±10.7	58.3±11.4*
Role emotional functioning of **REF**	81.8±7.5	60.5±9.4*
Mental Health **PZ**	81.1±9.5	59.6±11.0*
Psychological component of health, **PKZ**	78.9±5.7	55.8±5.7*

Note: SST - Shershevsky -Turner syndrome ; * significant decrease, $p<0.0001$

Low scores (less than 50 points) on the scales of social functioning (SF -32.6%), psychological health (27.9%) and vitality (25.6%) indicate a decrease in vital activity as a result of frequent fa-tigue of the respondents, limited social contacts, as well as the psychological distress of patients with SST.

Analysis of the psychological component of health revealed a decrease in the indicator by 29.3%. However, the lowest compared with clinical control were the indicators of VA (decrease by 32.6%) and SF (by 31.7%) , due to the psychological (decrease by 26.5%) and emotional state of patients (by 26.0%) (Fig.1b).

According to a number of researchers, Turner syndrome has a significant psychosocial impact on young girls and women. These effects may be associated with infertility, short stature, im-paired development of sexual characteristics, and, most importantly, lack of libido [11; 16].

Kesler S. [8] reads that shyness, anxiety, low self-esteem, and depression are often associated with awkwardness about appearance and/or infertility. There is evidence that adolescent girls with TSS are at high risk of developing problems associated with lower social activity, poor so-cial coping skills, and increased immaturity, hyperactivity, and impulsivity compared to their peers. In addition, patients with STS often have difficulty maintaining relationships with their peers [8; 10].

Findings

1.The main clinical signs of SST in the studied cohort were short stature (97.7%), primary amenorrhea (90.7%), sexual infantilism (79.1%) , a large proportion of patients have a wide chest (62.8%), X-shaped curvature of the legs (58.1%), skin folds on the neck (55.8%), more than a third were diagnosed with arterial hypertension (37.2%).

2.Almost half (48.8%) of patients with TSS have the classic form of TSS (monosomy X - 45, XO).

3.According to the results of the survey, there is a significant decrease in the parameters of the physical (by 26.5%) and psychological (29.3%) components of health and

quality of life.

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